

Questionnaire: Bethlem Myopathy

This questionnaire seeks to understand the connections between the mutated genes and constraints. It also aims to enable those affected to share information.

Genetic defect in:	COL6A1 <input type="checkbox"/>	COL6A2 <input type="checkbox"/>	COL6A3 <input type="checkbox"/>	
First symptoms appeared:	(e.g. at the age of 3 / 1980)			
Origin:	Inherited <input type="checkbox"/>	Spontaneous <input type="checkbox"/>	Unknown <input type="checkbox"/>	
Other family members affected / ill:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	What relationship? e.g. mother, son, grandchild	
Country of origin:				
Constraints				
Muscle weakness:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Contractures:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Hands:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Achilles tendon:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Cervical spine: Thoracic spine: Lumber spine:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Scoliosis:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Hamstring: (posterior thigh muscle)	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Jaw:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks: (e.g. constriction of the mouth opening, pain while chewing)	
Swallowing difficulties:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks: (e.g. only cold drinks)	
Heart:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	
Lungs:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks:	

Digestion:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks: (constipation, etc.)
Skin:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks: (e.g. eczema, poor scar healing)
Pain:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Where:
Secondary complications:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Type: (arthrosis etc.)
Medication:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Type:
Therapy:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Type: (physiotherapy, occupational therapy, heat treatment)
Results through therapy:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Detail:
Aids:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Type: (orthoses, crutches, wheelchair, etc.)
Support:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Type: (degree of care, household help, etc.)
Work capacity:	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Remarks: (part-time, retirement, etc.)
Negative effect on Bethlem myopathy?	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Detail: (overexertion of muscles, cold, stress, etc.)
Positive effect on Bethlem myopathy?	Yes <input type="checkbox"/>	No <input type="checkbox"/>	Detail: (rest, stretching, heat treatment, etc.)
Special features / remarks / symptoms not listed here:			

A sincere thank you for help in shaping this internet site:

www.bethlem-myopathie.de

No personal information such as name or personal details will be disclosed!
Best wishes, Diana Lauckner